Sickle Cell Anemia

Sickle cell anemia is a genetic disorder characterized by anemia, episodes of pain, and frequent infections. A person must have two copies of the HbS variant in the HBB gene in order to have this condition.

Overview Scientific Details

Jamie, you **have the HbS variant** we tested.

You could pass this variant on to your children.

How To Use This Test

This test does not diagnose any health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Carrier Status tutorial

See Scientific Details

Intended Uses

- To test for the HbS variant in the HBB gene.
- To identify carrier status for sickle cell anemia.
- Informs individuals with two copies of the HbS variant that people with their result are at risk of developing symptoms of sickle cell anemia.

Limitations

- This report does **not test** for other variants in the HBB gene.
- This report **only discusses** sickle cell anemia, not other forms of sickle cell disease. **See the Beta Thalassemia and Related Hemoglobinopathies report** for information about other variants in the HBB gene. In combination with the HbS variant, these genetic variants can cause other forms of sickle cell disease.
- For customers who purchased their 23andMe kit before November 2013, this test does **not report** if someone has two copies of the HbS variant.

Ethnicity Considerations

- This report is relevant for people of many ethnicities. It is most relevant for people of **African** descent, because the HbS variant is most common in people with African ancestry.
- In addition, because this report covers the only variant that causes sickle cell anemia, it is also relevant for other ethnicities in which the HbS variant is found. This includes people of **Middle Eastern** and **South Asian** descent, as well as people from the **Caribbean**, the **Mediterranean**, and parts of **Central** and **South America**.

You are a carrier.

You could pass this variant on to your children.

We detected one variant for sickle cell anemia.

People with only one variant are not expected to have sickle cell anemia. Instead, they are said to have sickle cell trait. **Learn more about sickle cell trait.**

Other variants in the HBB gene may also be relevant to you because they are associated with a related blood disorder. It's important to talk with a healthcare professional about additional testing. Your **Beta Thalassemia and Related Hemoglobinopathies** report may also be informative.

Your results may be relevant for you if you're thinking about starting a family.

If your partner is also a carrier for HbS or another variant in the HBB gene, each child may have a **25% chance** of having sickle cell anemia or a **related blood disorder**. Your relatives may also wish to consider testing, especially if they plan to have children.

About Sickle Cell Anemia

When symptoms develop

Symptoms typically develop by early childhood.

Typical signs and symptoms

- Anemia
- Fatigue
- Episodes of pain
- Frequent infections
- Stroke
- Injury to multiple organs

Ethnicities most affected

This condition can affect people around the world. It is most common in people of African descent. About 1 in 13 African Americans has the HbS variant. This variant is also found in people of Middle Eastern and South Asian descent, as well as people from the Caribbean, the Mediterranean, and parts of Central and South America.

How it's treated

Treatment focuses on managing pain and preventing complications. Certain medications or blood transfusions may improve symptoms. In addition, scientists are currently developing new treatment options that address the underlying cause of the condition.

Read more at: MedlinePlus GeneReviews Centers for Disease Control and Prevention National Heart, Lung, and Blood Institute

Consider talking to a healthcare professional.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Find a genetic counselor

Share your results with your family.

Share your report

To learn more about sickle cell trait and discuss options for additional testing, please talk to a healthcare professional.

Print report

Other variants in the HBB gene may also be relevant to you. See your Beta Thalassemia and Related Hemoglobinopathies report for more information.

See report

Learn more about this condition and connect with support groups.

Learn more

Sickle Cell Anemia

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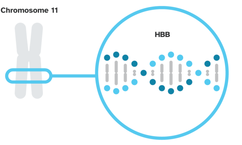
Overview | Scientific Details

Sickle cell anemia is caused by the HbS variant in the HBB gene.

HBB

The HBB gene contains instructions for making a protein called beta-globin. This protein is part of a larger protein called hemoglobin that is found in red blood cells. Hemoglobin transports oxygen from the lungs to all other cells of the body. Certain variants in the HBB gene alter the structure of hemoglobin, which can prevent it from transporting oxygen effectively.

Read more at MedlinePlus



You have one variant detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*	Additional Information
HbS **Gene:** HBB **Marker:** i3003137	**A** Variant copy from one of your parents **T** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [4, 5, 9, 11, 12, 13] | ClinVar

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation and Clinical Performance

Carrier frequency and carrier detection rate are most relevant for people without a variant detected.

For people who do not have the variant(s) tested, it may be possible to calculate an estimate of post-test carrier risk (the chances of still being a carrier) using information in this table. **View technical article on estimating post-test carrier risk.**

For people with one or more variants that could not be determined, their remaining chances of being a carrier may be similar to or less than the carrier frequency in people of their ethnicity.

Carrier frequency and carrier detection rate

This report provides two pieces of information to help interpret certain genetic results.

- **Carrier frequency** is the average chance of being a carrier for this condition. For example, a carrier frequency of 1 in 59 means that 1 out of every 59 people is expected to be a carrier for this condition.
- **Carrier detection rate** is an estimate of the percentage of carriers for this condition that would be identified by this test. For example, if the carrier detection rate is 80%, then our test is able to detect 80% of carriers for this condition. In cases where ranges are provided, the estimated carrier detection rate may depend on the region or country of ancestry.

Carrier frequency and carrier detection rate vary by ethnicity and are provided only where sufficient data is available.

Ethnicity	Carrier frequency	Carrier detection rate	References
Worldwide	Varies by ancestry	100% (This report covers the only variant that causes sickle cell anemia.)	[2 , 10]
African American	About 1 in 13	100%	[2 , 3]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Sickle Cell Anemia is indicated for the detection of the HbS variant in the HBB gene. This test is intended to be used to determine carrier status for sickle cell anemia in adults. This report also describes if a result is associated with personal risk of developing symptoms of sickle cell anemia, but it does not describe a person's overall risk of developing symptoms. The test is most relevant for people of African descent. It is also relevant for people of Middle Eastern and South Asian descent, as well as people from the Caribbean, the Mediterranean, and parts of Central and South America.

Special Considerations

- ACMG and ACOG recommend that people of all ethnicities who are considering having children should be offered carrier screening for hemoglobinopathies such as sickle cell anemia.

Test Performance Summary

Clinical Performance

This test includes the only variant that causes sickle cell anemia, so it is expected to detect all carriers. See the Test Interpretation and Clinical Performance section above for additional details about carrier detection rates.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the **Package Insert** for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. **American College of Obstetricians and Gynecologists. (2022). "Practice Advisory: Hemoglobinopathies in Pregnancy." Retrieved Oct 11, 2022, from https://www.acog.org/clinical/clinical-guidance/practice-advisory/articles/2022/08/hemoglobinopathies-in-pregnancy**
2. **Bender MA et al. (2003). "Sickle Cell Disease." [Accessed Oct 11, 2022].**
3. **Centers for Disease Control and Prevention. "Data & Statistics on Sickle Cell Disease." Retrieved Sep 16, 2022, from https://www.cdc.gov/ncbddd/sicklecell/data.html**
4. **Committee on Genetics. (2017). "Committee Opinion No. 691: Carrier Screening for Genetic Conditions." Obstet Gynecol. 129(3):e41-e55.**
5. **Darbari DS et al. (2013). "Severe painful vaso-occlusive crises and mortality in a contemporary adult sickle cell anemia cohort study." PLoS One. 8(11):e79223.**
6. **Gregg AR et al. (2021). "Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG)." Genet Med. 23(10):1793-1806.**
7. **Kato GJ et al. (2018). "Sickle cell disease." Nat Rev Dis Primers. 4:18010.**
8. **National Academies of Sciences, Engineering, and Medicine. (2020). "Addressing Sickle Cell Disease: A Strategic Plan and Blueprint for Action." National Academies Press.**
9. **Pawlowski JR et al. (2005). "Impaired vasodilation by red blood cells in sickle cell disease." Proc Natl Acad Sci U S A. 102(7):2531-6.**
10. **Piel FB et al. (2013). "Global epidemiology of sickle haemoglobin in neonates: a contemporary geostatistical model-based map and population estimates." Lancet. 381(9861):142-51.**

See all references

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
Jan. 25, 2023	The test can now report if a customer has two copies of the HbS variant. Customers with this result who purchased their 23andMe kit after November 2013 will see updated content in their report, including information about risk of developing symptoms of sickle cell anemia.
Dec. 9, 2019	The carrier frequency was updated for people with African American ancestry. Information in the report was generalized for people worldwide.
Feb. 18, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for this report may see an updated result.
Oct. 21, 2015	Sickle Cell Anemia report created.